

# **Pervasive Developmental Disorder (Autism)**

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# **Learning Objectives**

- **Define Autism.**
- **Delineate the clinical aspects of Autism.**
- **Recognize the Differential Diagnoses of Autism.**
- **Identify evaluation studies.**
- **Recognize treatment strategies for Autism and follow-up considerations.**

# **Autism: Definition**

**“A condition presenting in early childhood characterized by marked abnormalities in communication and social interactions and a restricted and socially atypical range of interests.”**

# **Autism: Definition**

- **Mental retardation**
  - **Moderate retardation**
  - **IQs generally fall between 35 and 50**
- **Seizure Disorders common**
  - **Movement disturbances**

# **Pathophysiology**

- **Findings vary in each individual**
- **Brain Abnormalities**
  - **Neuroanatomic, neuroimaging studies**
  - **Abnormalities of cellular configurations**
    - **Frontal lobe, temporal lobe, cerebellum**

# **Pathophysiology**

- **Abnormalities of cellular configurations (cont')**
  - **Enlarged cerebral areas**
  - **Amygdala, hippocampus**
- **Other whole body substances elevated**
  - **Serotonin, oxytocin, vasopressin**
- **Elevation of C-terminally directed beta-endorphin protein immunoreactivity**

# Pathophysiology

- May have altered metabolism of phenolic amines
  - Symptoms may worsen with certain foods
    - Dairy products
    - Chocolates
    - Corn
  - However, not proven
- Other hypotheses



# **Autism: Frequency**

- **U.S.**
  - **10-20 people per 10,000**
  - **Prevalence = 400,000 in U.S.**
  - **One of most common childhood developmental disorders**
    - **Epidemiological studies difficult**
      - **Tough to discern from different conditions (schizophrenia)**



# Autism: Frequency

- Internationally
  - 10-15% per 10,000
  - Higher in Japan



# **Autism: Frequency**

- **Race**
  - **More common in Japanese**
    - **Higher level of reporting?**
- **Sex**
  - **Male to female ratio = 3-4 : 1**
  - **Male children with normal karyotype (46XY)**
  - **10% have Fragile X syndrome**

# **Presentation**

- **Age**
  - **Early childhood**
  - **Absence of abnormalities in first 30 months highly unlikely to develop autism**

# **Presentation**

- **Age**
  - **Later presentation = other abnormalities**
    - **Childhood Disintegration**
    - **Rett Syndrome**
    - **Asperger Syndrome**
    - **Pervasive Developmental Disorder, NOS**

# **Autism**

## **Morbidity and Mortality**

- **Long-term outcome directly proportional to intelligence quotient (IQ) for each individual**

# Clinical

- **History**
- **Physical**
- **Causes**

# **Autism: History**

- **Proctodeclarative Pointing**
  - **Use of index finger to indicate item of interest to another person**
  - **Absence = predictive of Autism**
  - **Assessed by interview of family**

# History

- **Environmental Stimuli**
  - **Unusual responses to environment**
    - **Lack of a reaction or**
    - **Excessive reaction**
      - **To sounds, light, touch**





# **Autism: History**

- **Social Interactions**
  - **Separation from parent**
    - **Lack of appropriate eye contact**
    - **Indifference**
  - **Absence of typical response to pain**
  - **Trouble relating to others**

# **History**

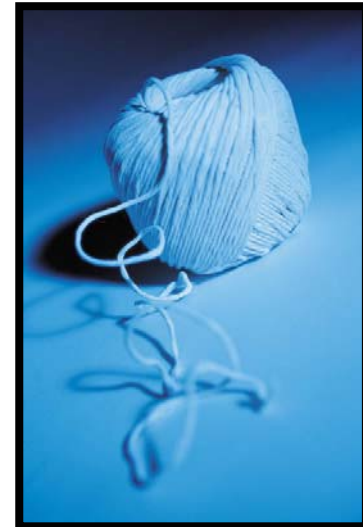
- **Specific Actions**
  - **Self injurious behavior**
  - **Aggression towards others**
  - **Temper tantrums**
  - **Hyperactivity**

# **Autism: History**

- **Communication**
  - **Speech abnormalities**
  - **Language delays**
  - **Language deviations**
  - **Pronomial reversals**

# History

- **Play**
  - **Absence of symbolic play**
  - **Nonfunctional Play**
  - **Repeated actions**
  - **Fascination of “non-toy” objects**



# **Autism: History**

- **Response to Febrile Illness**
  - **Decrease in odd behavior during illness**
  - **Ear infections, upper respiratory tract infections, fevers**
  - **Recovery from illness may coincide with a return of usual behaviors**

# **Autism: Physical**

- **Screening**
  - **CHAT (Baron-Cohen and colleagues)**
    - **Pretend Play**
    - **Normal Gaze monitoring**
    - **Proctodeclarative pointing**

# Physical

- **Screening**
  - **CHAT**
    - **Identify autistic toddlers in England**
    - **Reliability and validity not proved**
    - **Specificity and Sensitivity unknown**
    - **Possible cultural bias**

# **Autism: Physical Exam**

- **Screening**
  - **Body Movement**
    - **Choreoathetotic movements**
    - **Stereotypies**
    - **Motor tics**
    - **Hand Flapping**
    - **Spinning**



# Physical Exam

- **Screening**
  - **Body Features**
    - **Head Features**
      - **Elongated circumference**
    - **Palmer Crease**
      - **Single line across palms seen specifically in autistic children**



# **Autism: Physical**

- **Screening**
  - **Self-injurious Behavior**
    - **Skin picking**
    - **Self biting**
    - **Head or body punching or slapping**
    - **Lip chewing**
    - **Pulling out of hair or nails**

# **Autism: Causes**

- **Causes unknown**
  - **Hypotheses**
    - **“Refrigerator mothers”**
    - **Obstetric complications**
    - **Infections**
    - **Genetics**
    - **Toxic Exposure**

# **Autism: Causes**

- **Hypothesis**
  - **“Refrigerator mothers”**
    - **“Cold” parents**
    - **Emotionally distant from child**
    - **Hypothesis is that autism was adaptive for kids with uncaring parents**
    - **Disproven**

# **Autism: Differential Diagnoses**

# **Autism: Differential Diagnoses**

- **Acanthocytosis**
- **Anxiety Disorder: Obsessive-Compulsive Disorder**
- **Anxiety Disorder: Trichotillomania**
- **Biotin Deficiency**
- **Child Abuse & Neglect:**
  - **Dissociative Identity Disorder**
  - **Failure to Thrive**
  - **Physical Abuse**

# **Autism: Differential Diagnoses**

- **Child Abuse & Neglect: Psychosocial**
- **Dwarfism**
- **Child Abuse & Neglect: Reactive Attachment Disorder**
- **Cognitive Deficits**
- **Cornelia De Lange Syndrome**
- **Cri-du-chat Syndrome**
- **Down Syndrome**
- **Eating Disorder: Pica**

# **Autism: Differential Diagnoses**

- **Fragile X Syndrome**
- **Gaucher Disease**
- **Hearing Impairment**
- **Human Immunodeficiency Virus Infection**
- **Hypomelanosis of Ito**
- **Learning Disorder: Reading**
- **Toxicity - Lead**
- **Tuberous Sclerosis**



# **Autism: Differential Diagnoses**

- **44XXX Karyotype**
- **47 Chromosomes**
- **(7;20) Balanced chromosomal translocation**
- **Angelman Syndrome**
- **Deletion 1p35**

# **Autism: Differential Diagnoses**

- **Duplication of bands 15q11-13**
- **Extra bisatellite marker chromosome**
- **Habit disorder**
- **Hydrocephalus, infantile**

# **Autism: Differential Diagnoses**

- **Language disorder: mixed**
- **Language disorder: phonology**
- **Language disorder: receptive**
- **Language disorder: stuttering**
- **Long Y chromosome**

# **Autism: Differential Diagnoses**

- **Minamata disease**
- **Moebius syndrome**
- **Nonketotic hyperglycinemia (NKH)**
- **Partial 6p trisomy**

# **Autism: Differential Diagnoses**

- **Seizures**
- **Spasms, infantile**
- **Tourette disorder**
- **Trisomy 22**

# **Autism: Evaluation**

- **Lab Studies**
- **Imaging Studies**
- **Other Tests**

# **Autism: Evaluation**

- **Lab Studies**
  - **Whole Blood Serotonin**
    - **Elevated in 33%**
    - **Elevation occurs in parents and siblings**
  - **Serum Biotinidase**
    - **Reduced in autism**

# **Autism: Evaluation**

- **Lab Studies**
  - **Immunologic Studies**
    - **Decreased concentrations of C4B complement protein**
  - **C-terminally directed beta-endorphin protein immunoreactivity**
    - **Elevations common in autism**



# **Autism: Evaluation**

- **Radiological Studies**
  - **MRI: Results inconsistent but may show**
    - **Total brain enlargement**
    - **Lateral, fourth ventricle enlargement**
    - **Midbrain size reduction**

# **Autism: Evaluation**

- **MRI: Results inconsistent but may show (cont')**
  - **Reduction of medulla oblongata, cerebellar hemispheres, and vermal lobules VI and VII**
  - **Vermal hyperplasia/hypoplasia**

# **Autism: Evaluation**

- **Radiological Studies**
  - **CT: Also inconsistent**
    - **Can reveal**
      - **Specific deficits**
      - **Enlargement of ventricles**
      - **Hydrocephalus**
      - **Parenchymal lesions**

# **Autism: Evaluation**

- **Radiological Studies**
  - **Positron Emission Tomography (PET)**
    - **Reveals multiple deficits**
    - **No single finding universal to Autism**

# **Autism: Evaluation**

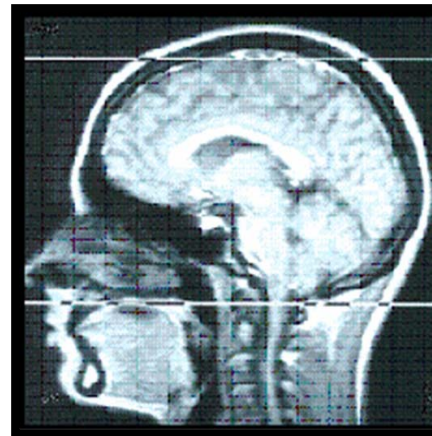
- **Single-photon emission computed tomography (SPECT)**
  - **Tests regional cerebral blood flow**
  - **Some have left hemisphere decrease**

# **Autism: Evaluation**

- **Radiological Studies**
  - **Electroencephalography**
    - **Rules out seizure disorder and related conditions**
    - **A single normal EEG does not rule out autism**

# **Autism: Evaluation**

- **Radioisotope Brain Imaging**
  - **Regional cerebral blood flow**
  - **Variable anomalies**



# **Autism: Evaluation**

- **Other Studies**
  - **Lead testing**
    - **Rule out lead poisoning**
- **Psychophysiological Assessment**
  - **Response habituation**
  - **Auditory overselectivity**



# **Autism: Treatment**

- **Medical Care**
- **Consultations**
- **Diet**
- **Activity**
- **Medications**

# **Autism: Treatment**

- **Medical Care**
  - **Individual Intensive Interventions**
    - **Behavioral**
    - **Educational**
    - **Psychological**
  - **Most effective treatments of disorder**

# **Autism: Treatment**

- **Medical Care**
  - **Starting care earlier demonstrates better outcome**
    - **Relies upon early diagnosis**
  - **Interventions on an individual basis**
  - **Weigh adverse effects of interventions against benefits**

# **Autism: Treatment**

- **Medical Care**
  - **Speech, behavior, and physical therapy**
    - **Facilitated Communication**
    - **Auditory Integration Training**
    - **Sensory Integration Therapy**
      - **Exercise and Physical Therapy**

# **Autism: Treatment**

- **Medical Care**

**“The most effective treatment for Autistic Disorder is special education . . . intensive individual special education by an educator familiar with instructing children with autistic disorder and related conditions.”**

# **Autism: Treatment**

- **Consultations**
  - **Metabolic**
  - **Immunologic**
  - **Otolaryngologic**
  - **Ophthalmologic**
  - **Neurologic**
  - **Neuropsychological**
  - **Infectious disease**

# **Autism: Treatment**

- **Diet**
  - **3 well balanced meals a day**
  - **Dietary consultation**
    - **Special diets may involve decreased gluten or casein**
  - **Vitamin supplements**
    - **B6 and Magnesium**



# **Autism: Treatment**

- **Activity**
  - **Exercise**
    - **Often therapeutic**
  - **Physical Therapist**
    - **May prescribe a specific exercise program**



# **Autism: Treatment**

- **Medications**
  - **Established treatments are nonpharmacologic**
  - **May be useful for comorbid conditions**
  - **High sensitivity possible with adverse effects**

# **Autism: Treatment**

- **Medications: Specifics**
  - **Serotonergic drugs**
    - **Examples: Naltrexone [generic], secretin**
    - **Reportedly beneficial**
    - **Six controlled clinical trials show no benefit**

# **Autism: Treatment**

- **Medications: Specifics (cont')**
  - **FDA allowing continued study of secretin**
  - **Danger of serotonergic syndrome**

# **Autism: Treatment**

- **Medications: Specifics**
  - **Ziprasidone [generic] *Geodon*®**
    - **Help control comorbid conditions**
    - **Controls aggression, irritability, agitation**

# **Autism: Treatment**

- **Medications: Specifics (cont')**
  - **Methylphenidate [generic] *Ritalin*®**
    - **Helps hyperactivity**
  - **Specific deficiencies**
    - **Biotin deficient individuals improve with biotin**

# **Autism: Follow-Up**

- **Complications**
- **Prognosis**
- **Education**
- **Medical / Legal Pitfalls**
- **Medications**

# **Autism: Follow-Up**

- **Complications**
  - **Physical Abuse**
    - **Vigilant physical exams recommended**
  - **Sexual Abuse**
    - **Maintain a high level of suspicion**

# **Autism: Follow-Up**

- **Prognosis**
  - **IQ highly correlated with prognosis**
  - **Low-functioning**
    - **May never live independently**
    - **Home or Residential Care**



# **Autism: Follow-Up**

- **Prognosis (cont')**
  - **High-functioning**
    - **Live independently**
    - **Hold jobs successfully**
    - **Marry and raise children**

# **Autism: Follow-Up**

- **Education**
  - **Family Education**
    - **Earlier diagnosis better**
    - **Individualized, intensive behavioral and psychological interventions**

# **Autism: Follow-Up**

- **Education**
  - **Legal consultation may be necessary to influence school board to fund appropriate education**

# **Autism: Follow-Up**

- **Education**
  - **School Education**
    - **Special Education performed by special educator shows the best outcome**
    - **School Boards may be ignorant of specific requirements**
      - **Seek information from knowledgeable sources**

# **Autism: Medical/Legal Pitfalls**

- **Delayed Diagnosis**
  - **Due to difficulty with diagnosis**
  - **Parents report concerns early to no avail at times**

# **Autism: Medical/Legal Pitfalls**

- **Treatment Issues**
  - **Effective treatment = Special Education**
  - **Education for All Handicapped Children Act of 1975**

# **Autism: Medical/Legal Pitfalls**

- **Obtaining Informed Consent**
  - **Highly vulnerable population**
  - **Patients, parents, guardians, surrogates must be made aware of diagnostic and treatment possibilities**
  - **Both written consent and videotape of the consent process**

# **Autism: Contacts**

**Autism Research Institute (ARI)  
4182 Adams Avenue  
San Diego, CA 92116**

**(619) 281-7165      Fax: (619) 563-6840**



# **Autism: Contacts**

**Autism Society of America [ASA]  
7910 Woodmont Avenue, Suite 650  
Bethesda, MD 20814-3015**

**(301) 657-0881, (800) 328-8476  
Fax: (301) 657-0869**

**Website: <http://www.autism-society.org/>**

# **Autism: Contacts**

**Cure Autism Now**

**5455 Wilshire Blvd., Suite 715**

**Los Angeles, CA 90036**

**1-323-549-0500 1-888-8AUTISM**

**E-mail: [info@cureautismnow.org](mailto:info@cureautismnow.org)**

**Web: <http://www.cureautismnow.org/>**

# **Autism: Contacts**

**OAR - Organization for Autism Research  
2111 Wilson Boulevard, Suite 600  
Arlington, VA 22201**

**(703) 351-5031**

**E-mail: OAR@autismorg.com**

**Web: <http://www.autismorg.com/>**

# **Autism: Contacts**

**Families for Early Autism Treatment**

**P.O. Box 255722**

**Sacramento, CA 95865-5722**

**(916) 843-1536**

**Web: <http://www.feat.org/>**

# **Autism: Contacts**

**MAAP (More Advanced Autistic People)  
Services Inc.**

**P.O. Box 524 Crown Point, IN 46307**

**Phone/Fax: (219) 662-1311**

# **Autism: Points to Remember**

- **Best outcome with early diagnosis**
- **Specific treatments improve outcome**
- **Affected individuals may range from low to high level functioning**
- **No specific tests as of now**
- **There is help for families**
- **Medico-legally a worrisome population**

# **Summary**

- **Definition of Autism**
- **Clinical Aspects**
- **Differential Diagnosis**
- **Treatment Strategies**
- **Follow-Up Considerations**